CTNND2 gene

catenin delta 2

Normal Function

The *CTNND2* gene provides instructions for making a protein called delta-catenin. This protein is active in the nervous system, where it likely helps cells stick together (cell adhesion) and plays a role in cell movement. In the developing brain, it may help guide nerve cells to their proper positions as part of a process known as neuronal migration.

In mature nerve cells, delta-catenin is located in specialized outgrowths called dendrites. Dendrites branch out from the cell and receive information from nearby nerve cells. This information is relayed across synapses, which are junctions between nerve cells where cell-to-cell communication occurs. Delta-catenin appears to play a crucial role in the function of synapses.

Health Conditions Related to Genetic Changes

cri-du-chat syndrome

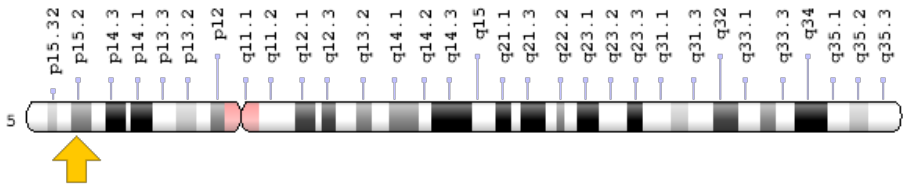
The *CTNND2* gene is located in a region of chromosome 5 that is often deleted in people with cri-du-chat syndrome. As a result of this deletion, many people with this condition are missing one copy of the *CTNND2* gene in each cell. The loss of this gene may cause severe intellectual disability in some affected individuals. Researchers suspect that intellectual disability could result from a disruption of neuronal migration during the early development of the nervous system.

People with cri-du-chat syndrome who do not have a deletion of the *CTNND2* gene tend to have milder intellectual disability or normal intelligence.

Chromosomal Location

Cytogenetic Location: 5p15.2, which is the short (p) arm of chromosome 5 at position 15.2

Molecular Location: base pairs 10,971,840 to 11,904,067 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- catenin (cadherin-associated protein), delta 2
- CTND2_HUMAN
- GT24
- neural plakophilin-related armadillo-repeat protein
- neurojungin
- NPRAP

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Neuronal Migration https://www.ncbi.nlm.nih.gov/books/NBK10831/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28CTNND2%5BTIAB%5D%29+OR+%28%28neural+plakophilin-related+armadillo-repeat+protein%5BTIAB%5D%29+OR+%28NPRAP%5BTIAB%5D%29+OR+%28neurojungin%5BTIAB%5D%29+OR+%28delta-catenin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CATENIN, DELTA-2
  http://omim.org/entry/604275

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CTNND2.html
- HGNC Gene Family: Armadillo repeat containing
  http://www.genenames.org/cgi-bin/genefamilies/set/409
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2516
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1501
- UniProt
  http://www.uniprot.org/uniprot/Q9UQB3

Sources for This Summary

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